Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: October 17, 2013

Subcommittee Members Attending:
Barbara Burton-Lurie Children's Hospital – Chair
Zohra Shad- University of Illinois at Chicago Hospital
Darrel Waggoner- University of Chicago
Lainie Friedman Ross – University of Chicago
Kathy Grange- Washington University- St. Louis
Tess Rhodes- Division of Specialized Services for Children

IDPH Staff:
George Dizikes, Arthur Kohrman, Khaja Basheeruddin, Rong Shao, Claudia Nash, Shannon Harrison, Jean Becker

Background
The meeting was called to order at 2:00 PM. Dr. Burton indicated that we will be modeling after the Missouri LSD screening experience and will also hold monthly meetings to review screening outcomes, which has been extremely beneficial in that state. Missouri has been screening for Krabbe since August 2012, and screening for Fabry, Gaucher, Pompe and MPS I since January 2013.

IDPH Laboratory Status Report
Dr. Dizikes reported that the lab has acquired four TQD instruments, with three being fully installed and the fourth partially installed in the newly remodeled space on the second floor of the IDPH Chicago lab. They have completed testing on 10,000 de-identified samples using the six-plex method of testing for Krabbe, Fabry, Gaucher, Niemann-Pick and MPS I using a three hour incubation period. They are successfully identifying confirmed positive samples that they have received. The substrate for MPS II testing may be available in the next few months. It is possible that testing for MPS II may be able to be incorporated into the existing six-plex assay if the incubation time is similar. The IDPH lab is awaiting further information from Dr. Michael Gelb of the University of Washington on this substrate.

The following timeline for implementation of LSD testing is being proposed by the IDPH lab:

February-March 2014: Finalize modifications to existing Perkin Elmer newborn screening database to prepare for LSD implementation.

February- March 2014: IDPH will monitor their testing effectiveness by conducting an exchange of 4,000 de-identified specimens with Perkin Elmer Genetics

May 2014: IDPH will pilot the screening on samples from one or two Illinois hospital to make sure the reports are being issued correctly, the database is functioning properly, etc.

June 2014: Statewide screening will be in place prior to July 2014.

Clinical Follow-Up Protocols

Dr. Burton indicated that she made the recommended changes to the disorder follow-up protocols that were suggested at the May meeting of this group. No further comments or suggestions were noted at this time.

Dr. Burton clarified that most females with Fabry will not be detected using an enzymatic method of newborn screening, and that the IDPH lab can add a disclaimer, or further clarification to their report if they desire to do so.

Dr. Burton also provided information on the outcome of newborns with Krabbe, in response to a question from Dr. Kohrman. She indicated that while stem cell transplant does prolong life, there is a 15%-25% mortality rate from the procedure and the outcome has not been as good in newborns diagnosed through screening as in siblings of affected children, most likely because of earlier diagnosis in siblings.

In New York, five newborns with Krabbe have been detected to date; 1 declined transplant and subsequently died, 4 received transplant. Of the 4 transplanted infants, 2 died of transplant complications, 1 is neurologically devastated and 1 has had severe ongoing transplant complications \but a reasonable neurologic outcome. Dr. Burton stated that it is imperative for physicians to explain these outcomes to parents prior to their decision about how to proceed with care once a diagnosis is made. She also indicated that only Lurie Children's Hospital, the University of Chicago and Washington University in St. Louis will be the centers which the IDPH Newborn Screening Program will recommend to physicians when reporting positive cases, since these are the only centers that can perform transplants.

Tess Rhodes of the Division of Specialized Care for Children (the Illinois Title V, Children with Special Health Care Needs Program) indicated they are committed to provide enzyme replacement therapy for children with Pompe disease. Pompe has been approved to be added to the Recommended Uniform Screening Panel (RUSP) by the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, but is awaiting final sign - off by Secretary Kathleen Sebelius.

Missouri Report

Dr. Grange reported the following from Missouri:

Disorder	~# Screened/Date Initiated	# Referred	# Diagnosed
Krabbe	100,000/August 2012	18	1 Krabbe, not infantile form, being monitored 1 suspected late onset Krabbe 9 carriers 1 parents refused testing; likely a carrier 3 genotype of unknown significance 3 pending diagnosis
Fabry	50,000/January 2013	53	16 confirmed Fabry 13 normal 1 refused follow up testing Other cases- pending results from centers

Gaucher	50,000/January 2013	14	1 confirmed Gaucher, mutations pending
Pompe	50,000/January 2013	31	2 early infantile Pompe 1 non-classical infantile onset 4 predicted late onset form 5 carriers 3 pseudodeficiency 9 normal 1 lost to follow up (moved away) 7 pending final results
Hurler	50,000/January 2013	39	1 confirmed Hurler 13 pseudodeficiency Remainder are normal or carriers

Dr. Grange reported that there will be a paper presented on pseudodeficiency genotypes at the upcoming meeting of the American Society of Human Genetics.

Next Steps

Dr. Burton indicated that since implementation of screening is not imminent, the next call should probably occur in two months, and was scheduled for Thursday, December 19 from 2-3 pm.

A data collection tool was discussed and Dr. Grange will share the spreadsheet used in Missouri, which can be emailed to the group for review. Missouri has created a separate spreadsheet for each disorder, to track all positive cases, which are identified by a unique reference number by center, and newborn screening lab ID number.

Prior to the December meeting of this group, Dr. Kohrman, Dr. Dizikes and Claudia Nash will discuss the possible creation of a database that can be accessible to the members of this work group and will draft a sample data collection tool, based on the Missouri model for the Illinois LSD work group to consider.